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Congenital Fetal Anomalies and the Role of Prenatal Ultrasound

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<http://dx.doi.org/10.5772/intechopen.71907>

Abstract

The ultrasound is the most widely used diagnostic tool in obstetrics nowadays, in particular in the detection of developmental disorders. However, it is important to know which are those disorders that can be detected prenatally with great certainty, and which ones can be detected only partially or not at all prior to giving birth. Pregnant women have high expectations, that any abnormalities should be fully recognizable and detected early during pregnancy, and this often leads to damages lawsuits. Thus, the right information is essential, so the doctors providing information also must have up to date knowledge about the effectiveness of ultrasound diagnostics. Prenatal diagnostics also entails enormous medical professional responsibility, since the consequences of an accidental inaccurate diagnosis can have significant consequences for both the fetus and the family. Thus, it is important to determine that how early and in what proportion the ultrasound protocol of the current Hungarian pregnancy care system is able to detect the individual disorder groups.

Keywords: congenital malformations, prenatal ultrasound, ultrasound detection

1. Introduction

Nowadays, 2D ultrasound is the most important diagnostic technique in obstetrics, especially in the diagnosis of congenital malformations.

The first diagnostic ultrasound screening usually takes place at around 11–13 weeks of pregnancy, when the thickness of the nuchal translucency is measured and the presence of the nasal bone is confirmed. Nuchal translucency (NT) is an excess fluid under the nuchal skin of the fetus in the first trimester. In 1866, Langdon Down described this phenomenon as trisomy 21 patients' having "too large skin" [1]. In the 1990s it was recognized that the excessive fluid

is responsible for this thickening and that it was possible to measure the thickness of this fluid at the back of the neck around the third month of pregnancy [2, 3]. A thicker nuchal translucency might suggest chromosomal abnormality, but it can be present in cardiovascular malformations and genetic syndromes as well.

NT is usually measured around 11–13 weeks of gestation. The thickness of the nuchal oedema is proportional to the crown-rump length (CRL) of the fetus so it is measured when the CRL is 45–84 mm. The higher the NT is at a specific CRL, the higher the risk for chromosomal abnormalities is [2]. For example, in Turner-syndrome, the NT value is around 8 mm higher than the median [2].

In Down-syndrome, nasal bone is absent in 60–70% of the affected fetuses. Langdon Down observed that those with 21-trisomy had small noses, caused by the hypoplasia of the nasal bone. The hypoplasia of the nasal bone can also be detected during pregnancy. According to a meta-analysis, the nasal bone was absent in 1.4% of healthy fetuses, while in 69% of fetuses with trisomy 21. Furthermore, the maxilla was shorter in 25% of the affected fetuses, and ductus venosus flow was abnormal in 80% [1, 2].

In Edwards-syndrome (trisomy 18), early growth retardation and bradycardia can be detected at 11–13 weeks. Also, the nasal bone is absent in 55% of the cases and 75% has a singular artery in the umbilical cord.

In Patau syndrome (trisomy 13), 70% of the fetuses have tachycardia. Early growth retardation, megacystis and holoprosencephaly are also frequently present [2].

The second ultrasound screening is usually done around 18–20 weeks of gestation. The goal of this screening is to diagnose congenital malformations and to detect other signs of chromosome anomalies and other syndromes. Therefore, this ultrasound examination is called “genetic screening.”

Structures to be examined:

- cranium (BPD, occipito-frontal diameter-OFD, head circumference-HC)
- face
- spine (spina bifida)
- heart
- diaphragm
- stomach (filling of the stomach)
- abdominal wall (anteroposterior and transversal diameter-AD, abdominal circumference-AC)
- kidneys, bladder
- extremities (femur length-FL, humerus length-HL)
- placenta, umbilical cord
- amniotic fluid
- uterine artery Doppler

Malformations are usually easier to diagnose later in pregnancy as the organ develops and grows (i.e., heart malformations). Also, it is easier to detect anomalies when the defect grows with the gestational age (i.e., pyelectasis).

2. Central nervous system

Brain anomalies are one of the most common group of congenital malformations. To be examined: the cerebellum, choroid plexuses, cisterna magna, lateral ventricles, cavum septi pellucidi. There are three major scan planes for the fetal brain:

1. thalamic view: BPD, HC measurement, thalamus, cavum septi pellucidi
2. ventricular view: lateral ventricles, choroid plexuses, arteria
3. cerebellar view: cerebellum, cisterna magna (Between the 15–22 weeks of gestation, the diameter of the cerebellum in mms is usually equal to the gestational weeks of the pregnancy.) [3, 4]

Any abnormality in these views may suggest a brain malformation, such as: neural tube defects, ventriculomegaly, holoprosencephaly, hydranencephaly, Dandy-Walker malformation, agenesis of the corpus callosum, porencephaly, intracranial tumor. The sensitivity of the ultrasound is high in the diagnosis of these malformations [5, 6].

Neural tube defects (NTDs) are the second most common types of malformations. The incidence of NTDs is around 1:1000 in the USA and 8:1000 in the UK. Anencephaly, encephalocele/meningocele and spina bifida are the most common NTDs [7–9].

Anencephaly is the absence of a major portion of the skull and the brain hemispheres caused by an abnormal closure in the cranial part of the brain. Exencephaly is an early stage of anencephaly, when the brain is still present but is located outside the skull. Anencephaly develops as the neural tissue degenerates. Exencephaly can be detected by first trimester ultrasound as floating brain tissue outside of the skull. CRL is usually lower than normal. In the second and third trimester, polyhydramnios is usually also present. Anencephaly is a fatal condition and most affected pregnancies are terminated after the diagnosis [7, 8].

Encephalocele and meningocele are defects of the skull. In the former, brain tissue and meninges protrude through the defect of the skull, while in the latter only the meninges are affected. The defect of the skull can be detected with ultrasound. In 75% of the cases the defect is occipital.

Spina bifida is present when the spine does not close properly. There are three types based on what structures are affected:

1. occulta: no protrusion, covered with skin
2. cystica: protrusion, covered or not covered (**Figure 1**)
 - a. meningocele: contains meninges and cerebrospinal fluid
 - b. myelomeningocele: contains meninges, cerebrospinal fluid and neural structures
3. rachischisis/myeloschisis: the neural tube is completely open, no meninges or skin

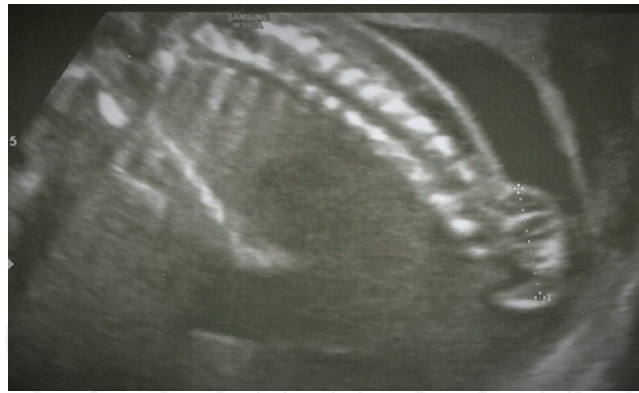


Figure 1. Spina bifida cystic. The spine does not close properly. The cystic protrusion contains meninges and cerebrospinal fluid.

The ultrasound detection rate of spina bifida is almost 100% thanks to the specific markers: banana and lemon signs (**Figure 2**), small posterior fossa, small cerebellum, and ventriculomegaly. Also, there is a V-shaped appearance to the posterior elements of the spine [10, 11].

Around 88% of all NTDs can be detected with prenatal ultrasound (25–94%) [12]. More than 90% of anencephaly and encephalocele cases are detected, while only 44% of fetuses with spina bifida are diagnosed in the first trimester [13–15]. However, the efficacy of ultrasound in the detection of spina bifida is 92–95% in the second trimester [12, 16–18].

In ventriculomegaly, the ventricles are dilated by the cerebrospinal fluid. Fluid accumulation in the ventricles is caused by excessive production, abnormal absorption or impaired circulation. The pressure increases in the ventricles and compresses and ultimately damages the brain tissue (internal hydrocephaly). When the amount of cerebrospinal fluid increases in the subarachnoid space, we talk about external hydrocephaly. Macrocephaly is when the skull is dilated too. The first sign on the ultrasound is usually the dilation of the occipital horn of the lateral ventricles. We talk about ventriculomegaly, when the dilation is larger than 8 mm at 18 weeks of gestation. Later in pregnancy, ventriculomegaly is present when the lateral ventricle/hemisphere ratio is higher than 0.5 (**Figure 3**) [4, 5]. When measured in the atrium (the connecting point of the occipital and temporal horns), a 10–15 mm dilation is mild, while



Figure 2. Lemon sign. In cases of spina bifida, special signs like lemon sign are visible in some cases.



Figure 3. Agenesis/dysgenesis of corpus callosum. The teardrop shaped lateral ventricles, in some cases absence of a normal cavum septi pellucidi, dilatation of the third ventricle may suggest this malformation.

a >15 mm dilation is severe. Furthermore, BPD is increased in most cases. After the detection of ventriculomegaly, toxoplasma and virus serology should be considered. When present with other anomalies, chromosomal examination is also due. The postnatal management is shunt implantation in most cases [4, 5, 11].

Holoprosencephaly is present when the prosencephalon (forebrain) of the embryo fails to divide into two hemispheres. This causes a sequence of malformations: cyclopia, cyclotia, ethmocephalia, proboscis, cebocephalia, premaxillary agenesis, cheilognathopalatoschisis.

Types of holoprosencephaly:

1. alobar: the longitudinal fissure, the falx cerebri, the septum pellucidum and the corpus callosum are absent
2. semilobar: the frontoparietal part of the brain is undivided
3. lobar: hemispheres are separated, absence of the septum pellucidum, fusion of the lateral ventricles
4. arhinencephaly: absence or hypoplasia of the olfactory tract and bulb

Birth prevalence is around 1:10,000–1:15,000, but the prevalence is much higher in miscarriages. Chromosome abnormality is associated in half of the cases, most frequently 13-trisomy. The alobar type is fatal, while patients with lobar or semilobar holoprosencephaly suffer from severe physical and mental disabilities [4, 5, 18].

Agenesis of the corpus callosum (ACC) (**Figure 3**) is among the most frequent malformations of the developing brain with an incidence of around 5:1000 [8, 11]. Corpus callosum can be visualized at the end of the first trimester using the Doppler flow technic [2]. The absence of a normal cavum septi pellucidi, teardrop shaped lateral ventricles, colpocephaly and dilatation of the third ventricle may suggest ACC [4, 11].

Dandy-Walker malformation consists of hydrocephaly, hypoplasia/agenesis of the cerebellar vermis and dilatation of the fourth ventricle (cyst of the posterior fossa). The birth prevalence is 1:12,000. Dilation of the cisterna magna and the fourth ventricle can be seen on the ultrasound [5, 8].

Based on our findings, 255 out of 351 congenital craniospinal malformations were diagnosed prenatally (72.65%).

We found that the sensitivity of ultrasound was high in case of the anencephaly/exencephaly (95%), spina bifida (88.89%), hydantencephaly (87.5%) and ventriculomegaly (80%) groups. However, the sensitivity of ultrasound was lower in the corpus callosum agenesis (50%), microcephaly (25%) and Arnold-Chiari malformation groups.

3. Face

Malformations of the facial structures are often minor anomalies, therefore, they do not cause any functional impairment. However, they can suggest chromosomal abnormalities or other, more severe congenital anomalies (malformations of the heart or brain) so diagnosing these minor anomalies is important.

Facial structures to be examined are the nose, lips, ears and chin. The efficacy of prenatal ultrasound is low in case of these malformations and they are often left undiagnosed. As ultrasound techniques develop, the detection of anomalies such as cleft lip and palate (**Figure 4**) before birth is getting more accurate, reaching a sensitivity of 14–25% [19, 20]. 3D technique proved to be more effective in assessing the facial structures of the fetus [21].



Figure 4. Cleft lip. The detection of anomalies such as cleft lip and palate before birth is getting more accurate.

In our study, only 43 out of 135 face malformations were diagnosed prenatally (31.85%). We found that cleft lip and palate was detected with the highest sensitivity (53.33%), while no choanal atresia cases and only 9.09% of micrognathia cases were found.

4. Cardiovascular system

Congenital malformations of the cardiovascular system are the most common malformations with a birth prevalence of 8:1000. Half of these malformations are severe and life-threatening

[18, 22, 23]. 30–40% of these anomalies occur in association with other malformations or chromosomal abnormalities [24, 25].

Risk factors are abnormal NT, heart malformation in the mother's history (mother, family, previous pregnancies), diabetes of the mother, heart rate anomalies in early pregnancy, teratogenic exposure in early pregnancy. The presence of even one of these factors means that the pregnancy is high risk for congenital heart malformations [5, 23, 26].

Low-risk pregnant women go through a routine screening ultrasound at 18–22 weeks. During routine screening both the four-chamber and the two outflow tract views are to be examined. The four-chamber view can be visualized in a transversal plane above the diaphragm. The size of the fetal heart, its location, rhythm, the cardiac axis, the two atriums, the two ventricles, the ventricular septum, the atrial septum primum and the atrioventricular valves can be examined in this view [27]. Around 60% of major cardiovascular malformations can be detected in the four-chamber plane. The outflow tract views (left and right) give information about the anatomy of the aorta, pulmonary artery, aortic and pulmonary valves and the origin of the aorta and pulmonary arteries. They can be visualized by sliding the transducer toward the fetal head from a four-chamber view [27]. The detection rate of cardiovascular anomalies was higher when the four-chamber and outflow tract views were all examined [5, 26, 27].

In the high-risk group, an early fetal echocardiography is performed. Fetal echocardiography can be done from 11 weeks of gestation, and almost all of the malformations can be detected by 14 weeks (84–95%) [28, 29]. The examination can be repeated around 20 weeks. Cardiomyopathies, valvular stenosis, and tumors can only be detected later in pregnancy [5, 30].

In our study, 67.7% of all cardiovascular malformations were diagnosed with ultrasound. We found high ultrasound sensitivity in the univentricular heart (96.43%), pericardial effusion (90.91%) and hypoplastic left heart syndrome (90%) groups. Though, atrial septum defect and pulmonary artery malposition cases were detected with the lowest sensitivity (31.71% and 33.33%).

5. Lungs, diaphragm

Malformations of the lung are rare anomalies, but diagnosing them prenatally is still important, especially to determine appropriate postnatal management. At 18 weeks of pregnancy, the lungs can be visualized around the heart, filling two-third of the thorax. The quantity of the amniotic fluid has an important role in the development of the lungs. Therefore, in severe oligohydramnios the lungs become hypoplastic (Potter-sequence).

Cystic malformations of the lungs can be separated into three groups: solitary and multiplex cystic anomalies and congenital cystic adenomatoid malformation (CCAM). The latter is a multicystic hamartoma that is usually confined to only one lobe. 47–80% of all lung malformations are CCAMs with a prevalence of 0.3–0.9/10,000 [31–33]. Ultrasound detection depends on the size of the cyst: Type I: 10–20 mm, Type II: 5–10 mm, Type III: small, not detectable.

If the anomaly is large it may dislocate the mediastinal structures, causing polyhydramnios through the compression of the esophagus. It is often associated with other malformations such as cardiovascular, urogenital, and skeletal anomalies or hydrocephalus.

In pulmonary sequestration, the hamartoma usually gets its blood flow from the systemic circulation, either from the abdominal or the thoracic aorta. Most often small cysts (Type III) can be seen on the ultrasound, but it is important to prove the hamartomas connection to the systemic circulation. The intralobular form is more prevalent, but is less likely to get diagnosed. 90% of the extralobular cases are located in the left lower lobe and it is easier to detect [33, 34].

In diaphragmatic hernia, abdominal organs get through a defect of the diaphragm to the thorax. The incidence of this malformation is around 1:3700. 90% of the hernias are present on the left side. The defect itself, abdominal organs in the thorax and mediastinal shift can be detected with ultrasound. Pulmonary hypoplasia can also develop in severe cases due to the volume expansion in the thorax [5, 35].

Malformations of the lungs were detected with a 52.94% sensitivity in our study, while diaphragmatic hernia cases were diagnosed with ultrasound in 86.79% of the cases.

6. Abdominal wall

Abdominal wall malformations (omphalocele, gastroschisis) are fairly prevalent malformations. Maternal serum alpha-fetoprotein level is often elevated in these malformations and intrauterine growth restriction (IUGR) is frequently present. IUGR and the involvement of the liver are important predictive factors for the outcome of these pregnancies [36, 37].

In gastroschisis, there is a defect on the abdominal wall that affects all the abdominal layers, including the amnioperitoneal membrane. It usually appears on the right side of the umbilical cord, but does not involve the cord itself. Gastroschisis has an incidence of 1:2000–1:5000 and is more prevalent in the fetuses of younger mothers. Gastroschisis is always associated with polyhydramnios. The efficacy of ultrasound in this anomaly is around 80% at 18–20 weeks [38, 39]. In early diagnosis, termination of pregnancy is an option and when diagnosed later, it is important to follow-up on the condition of the intestines and deliver the baby if signs of necrosis appear. Cesarean section is suggested in all cases, because vaginal delivery pose a higher risk of infection of the abdominal organs. The fetus is delivered before 35 weeks of pregnancy, because the chance of a successful reposition of the organs is lower afterwards [5, 36, 40].

In omphalocele, abdominal organs herniate into the amniotic fluid through the umbilicus. The defect is always associated with polyhydramnios, it is medially positioned and the organs are covered by the amnioperitoneal membrane. Omphalocele has an incidence of 1:6000 live birth. Herniation of the abdominal organs to the umbilical cord is normal before 11 weeks, but the defect usually closes by then. Therefore, omphalocele can be only detected with second trimester ultrasound at 18–20 weeks. The sensitivity of prenatal ultrasound in the diagnosis of this anomaly is around 75–90% [6, 7, 38, 39]. Performing echocardiography or cytogenetic

examination is justified in these fetuses as omphalocele is associated with other malformations and chromosomal abnormalities in more than half of the cases. The smaller the defect is, the higher the risk of aneuploidy is. When there is no associated malformation, the pregnancy can be carried to term [40].

Abdominal wall malformations were diagnosed with a high sensitivity in our study. All gastroschisis (12/12) and most of omphalocele (25/33, 75.76%) cases were diagnosed antenatally.

7. Gastrointestinal system

The fetal stomach can be visualized with ultrasound after 14 weeks. During the second and third trimester, the liver, gall bladder, spleen and intestines can all be examined with ultrasound in most cases. When the stomach cannot be seen it may suggest malformations such as esophageal atresia, diaphragmatic hernia, abdominal wall anomalies or neurological problems. When the filling of the stomach is not visualized during the examination, the ultrasound has to be repeated [5, 36].

Esophageal atresia is the absence of a part of the esophagus. The atresia is positioned higher than the trachea bifurcation in 85%, and a tracheoesophageal fistula is present in 90% of the cases. The birth prevalence of this malformation is around 1:3000. Signs on the ultrasound are polyhydramnios and the absence of the filling of the stomach. However, when a fistula is present, the stomach is filling, hence the low prenatal detection rate (10–40%) and late, third trimester diagnosis [14, 16, 34]. Later on, at around 24 weeks of pregnancy, the dilatation of the proximal end may be seen. About half of the cases are associated with other malformations, aneuploidy in 20%, growth retardation in 40% and most often with cardiovascular anomalies. Therefore, performing echocardiography and cytogenetic examination is important [5, 36, 41, 42].

The appearance of the intestines changes with the development of the fetus. Increased echogenicity of the fetal intestines can be a normal variant, but can also appear after the ingestion of blood. An increased echogenicity (as high as the bones) may appear in gastrointestinal malformations, Down-syndrome, cystic fibrosis or congenital infections (such as cytomegalovirus) [5, 36, 43].

Duodenal atresia may occur due to a real atresia, membranous closure or compression (annular pancreas) of the duodenum. The incidence of this condition is around 1:6000–1:10,000 birth. One-third of the cases are associated with Trisomy 21 and 50% develop as part of multiplex malformations. The specific ultrasound finding for duodenal atresia is the “double-bubble” sign. The two bubbles are the distended stomach and proximal duodenum. Usually polyhydramnios also appears. Echocardiography and cytogenetic examination is needed to detect the associated anomalies [5, 36].

Intestinal atresia only affects the small intestines in 95% of the cases with an incidence of 1:10,000 live birth.

Morphological types:

- Type 1: The intestines are intact after the atresia
- Type 2: There is a narrowing after the atresia and the intestines are often shortened
- Type 3: multiplex anomalies of the intestines
- Type 4: The dorsal mesentery is absent and the intestines are shortened

Intestinal atresia mostly occurs due to teratogenic effects. Polyhydramnios and dilated intestinal loops are usually seen on ultrasound. Atresia of the large intestines and of the anus is harder to detect due to the lack of polyhydramnios and less distended intestines. The dilated rectum that is filled with water may be visualized between the sacrum and the bladder [5, 36, 43].

Hirschsprung's disease is a congenital aganglionosis of the intestines that causes the distal large intestines to dilate. It occurs in 1:5000 live birth, mostly in boys. Dilated intestines and polyhydramnios are the most important signs on ultrasound after the second trimester. However, it is hard to differentiate between Hirschsprung's, cystic fibrosis and the atresia of the large intestines based on the ultrasound findings [5].

In our study, duodenum atresia was diagnosed with a high sensitivity of 94.74%, while atresia of the esophagus was diagnosed in only one-fifth of the cases.

8. Urogenital tract

Urogenital malformations are the most often diagnosed anomalies with a birth prevalence of 0.5%. Fetal kidneys can be first visualized with ultrasound at 14 weeks of pregnancy beside the spine and by 18 weeks, their structure can be analyzed too [44].

In the first 18 weeks of development, the amniotic fluid is derived from the placenta and membranes, but after 16 weeks, fetal kidneys gradually take over the production. Therefore, anomalies of the fetal urinary tract may result in impaired production of the amniotic fluid and eventually oligohydramnios. However, when one kidney is functional, the quantity of the amniotic fluid can be normal as well [44].

Renal agenesis is the absence of one or both kidneys. The birth prevalence of this abnormality is 1:4000. The fetal kidney or kidneys cannot be visualized with ultrasound, only the enlarged adrenal glands (lying down adrenal sign). When both kidneys are absent, the filling of the bladder is missing and oligohydramnios is severe. The severe oligohydramnios may cause Potter-syndrome: flattened nose and ears, peculiar look, hip dysplasia, club-foot, hypomelia, sirenomelia, arthrogryposis, fetal growth retardation and pulmonary hypoplasia (due to the impaired secretion and resorption of the amniotic fluid and compression) [5, 44–46].

Potter type I polycystic kidney is usually bilateral and shows an autosomal recessive inheritance pattern. It occurs in 1:10,000–1:40,000 birth. The cysts are small, 1–2 mm, originated from the collecting ducts. Cysts can also appear in the liver and renal and hepatic fibrosis may also

occur. The cysts are too small to be detected with ultrasound, but the enlargement and hyper-echogenicity of the kidneys are seen. Furthermore, oligohydramnios and the absence of the filling of the bladder are also usually present. Potter-sequence may also appear due to the severe oligohydramnios [5, 44, 46].

In multicystic renal dysplasia, there is no normal parenchyma, but 10–20 mm cysts and connective tissue fill the kidneys. The anomaly is unilateral in two-third of the cases. The quantity of the amniotic fluid is usually normal, but oligohydramnios can appear in bilateral and sometimes polyhydramnios in unilateral cases. The incidence of this malformation is around 1:10,000 birth and it is more prevalent in boys. Most cases are diagnosed with ultrasound at 18 weeks as the cysts can be visualized and they are not connected to the renal pelvis. Also, the kidneys usually have an abnormal shape. Bilateral dysplasia is often fatal [44–46].

Obstructions of the urinary tract usually results in the dilatation proximally. Obstruction of the ureteropelvic junction is the main cause of hydronephrosis in the neonate. Pyelectasis is the dilatation of only the renal pelvis and the calyces, while when the parenchyma is also affected by the compression, we talk about hydronephrosis. The dilatation of the pyelon and calyces can be seen in pyelectasis, while hydronephrosis appears as a solid cyst or sac. Pyelectasis is defined as a pyelon larger than 4 mm before 20 weeks or larger than 7 mm after 34 weeks of gestation. The anomaly is often diagnosed (2–5.5% of all fetuses), but spontaneous regression is common [6, 7, 45, 47].

In the obstruction of the ureterovesical junction, the ureter distends as well, creating a megalo-ureter. This anomaly is 4 times more prevalent in boys. In the presence of a posterior urethral valve, the bladder, ureter and renal pelvises dilate too and the distention damages the kidneys as well. It may cause severe oligohydramnios and Potter-sequence. Also, the dilated organs may stretch the abdominal wall, causing rectus diastasis (prune belly syndrome) [5, 44, 45].

Ovarian cyst is the most common abdominal mass in female fetuses with a birth prevalence of 1:2600. The malformation is more and more often diagnosed prenatally. The etiology of the anomaly is unknown and the cyst is usually benign. Ovarian cysts are more prevalent in fetuses of mothers with diabetes, eclampsia or Rh isoimmunisation. Complications are rare: compression of other organs, rupture, bleeding. The most common complication is the torsion of the cyst may cause the ischemia and eventually necrosis of the ovary [47, 48].

Malformations of the female organs are mostly caused by the impaired development of the Müllerian duct. When the two ducts do not fuse properly it causes the female organs to be septate or doubled. MRKH (Mayer-Rokitansky-Küstner-Hauser) syndrome is a malformation of the Müllerian duct when the upper two-third of the vagina and the uterus are missing [49].

Malformations of the urogenital tract were diagnosed with ultrasound in 54.55% of the cases. The sensitivity of the ultrasound was high in polycystic kidney (100%), obstructions of the urinary tract (88.89%), multicystic renal dysplasia (80.57%), and pyelectasis (67.21%). However, genital malformations were harder to diagnose and a correct diagnosis was made in only 19.7% of the cases.

9. Extremities

Congenital malformations of the extremities may appear as solitaire anomalies or as multiplex abnormalities associated with syndromes. Most of these malformations are hard to diagnose prenatally, the sensitivity of ultrasound is around 25% [50, 51]. Measurements of the length of the femur and the humerus are part of the fetal biometry [52, 53].

Club-foot (**Figure 5**) is the most prevalent congenital malformation of the extremities with a birth prevalence of 1:1000. However, according to some studies, the malformation occurs in 1:250 in utero [54]. The affected foot is rotated internally. In half of the cases, both feet are affected and club-foot is associated with other abnormalities (such as Trisomy 18). Also, it may occur as part of the Potter-sequence, or in neuromuscular anomalies, neural tube defects or amniotic band constriction [54].



Figure 5. Club-foot. On the picture the affected foot is rotated internally.

In our study, malformations of the skeleton were diagnosed with a higher sensitivity than anomalies of the extremities (82.93% vs. 37.5%). We found higher ultrasound sensitivity in osteogenesis imperfecta (80%), reduction deformities (64.71%), and club foot (51.43%). Ultrasound was less effective in diagnosing hip dysplasia and malformations of the fingers.

10. Placenta, umbilical cord

The placenta is the organ that connects the mother to the fetus and ensures the normal development and growth of the fetus. It is important to examine the location (especially the relative position to the cervix) and morphology of the placenta [55].

Placenta praevia is an anomaly when the placenta is inserted either partially or totally in the lower, passive segment of the uterus. It can only be diagnosed after the second trimester because the site of insertion usually shifts upwards with the growth of the uterus. Placenta praevia has a birth prevalence of 2.8:1000 and it is more common in twin pregnancies and after a previous cesarean section [56, 57].

When the placenta is abnormally attached, it may reach the myometrium (placenta accreta), the serosa (placenta increta) or other organs (placenta percreta). The detection of placenta accreta is hard, but increta and percreta are easier to visualize. The birth prevalence of placenta accreta is 1:2500, while in placenta praevia cases, the prevalence is 1:10 [56, 57].

The maturity of the placenta was classified by Grannum in 1988 based on the ultrasound image:

- Grade 0: First two trimesters, the chorionic plate is smooth, uniform echogenicity
- Grade I: 18–29 weeks, indentations of the chorionic plate, occasional echodensities
- Grade II: 30–36 weeks, deeper indentations, echodensities
- Grade III: after 36 weeks, complete indentations, large echodense areas, calcification

When the maturity of the placenta and the fetal biometry are discordant, it suggests intrauterine growth retardation [55, 57, 58].

Examination of the insertion site of the umbilical cord is also important as it provides the nutrient supply going to the fetus. Marginal insertion is often associated with intrauterine growth retardation. In case of velamentous cord insertion, the umbilical vessels are only covered by the amniotic membrane, therefore they are less protected. Also, this anomaly is often associated with the presence of a single umbilical artery [57]. Single umbilical artery is present in 0.2–1% of the pregnancies. It is a minor anomaly that is often associated with cardiovascular, brain and urogenital malformations and Trisomy 13 or 18 [57, 58].

Normally, the umbilical cord is 50 cm long at terminus. When it is shorter than 30 cm, it is classified as short, while a long umbilical cord is over 80 cm. The length of the cord affects the mobility of the fetus, therefore it is important to examine this feature. Furthermore, the degree of coiling of the umbilical cord should be determined (CI: coiling index) as the absence of coiling may suggest chromosomal abnormality, fetal distress or retardation [57, 58].

11. Amniotic fluid

The amniotic fluid is made by the placenta and the membranes before 16–18 weeks, while after 16 weeks, fetal kidneys gradually takes over the production up until birth. Kidneys excrete around 5 ml/h of urine after 20 weeks, which increases to 50 ml/h by the end of pregnancy. Abnormal quantity of the amniotic fluid may indicate a congenital malformation or chromosomal abnormality. Amniotic fluid index (AFI) is an objective method for determining the amount of the amniotic fluid. In the “four quadrant technique,” the vertical length of each pocket of fluid is measured in each of the four quadrants and then the measurements are summarized. A normal AFI is 8–24 cm after 16 weeks of gestation. The other technique is the “single deepest pocket” technique measures the vertical length of the deepest pocket with a normal value of 2–8 cm. The latter is mostly used in twin pregnancies [57, 58].

Oligohydramnios is the condition when there is less amniotic fluid than the normal (less than 500 ml in the third trimester).

Types based on pathophysiology:

- Amniotic: premature rupture of the membranes
- Maternal: smoking, fasting, low fluid intake
- Fetal: urogenital malformations
- Fetomaternal/placental

The fundus height is lower than normal and the fetal movements are dim, often painful. The AFI is less than 7 cm with the “four quadrant technique” and less than 2 cm measured in the deepest quadrant. Anhydramnios is the condition when the amniotic fluid is missing. Severe oligohydramnios may result in Potter sequence in the fetus. When oligohydramnios is caused by bilateral renal agenesis, the condition is called Potter-syndrome. In the third trimester, uterine contractions may result in the compression of the placenta and umbilical cord, endangering the fetus. Prenatal mortality in oligohydramnios is around 10% [58].

Polyhydramnios is an excess of amniotic fluid around the fetus, more than 2000 ml in the third trimester. It occurs in 1–2% of pregnancies.

Types based on pathophysiology:

- Amniotic: infection, chorioamnionitis
- Maternal: diabetes mellitus, preeclampsia, pyelonephritis, syphilis
- Fetal: twin pregnancy, congenital anomalies affecting the swallowing or resorption of the amniotic fluid
- Unknown

The fundus height is usually bigger than normal, the abdomen is large and tense. Mothers usually have dyspnea and the fetal heart sounds are faint. On ultrasound, a large echoless space can be seen between the fetus and the uterine wall with the umbilical cord freely floating. The polyhydramnios is mild when the vertical measurement of the deepest pocket is 8–11 cm, and severe when it is over 16 cm. AFI is usually more than 24 cm in the four quadrants. Cytogenetic examination is needed when polyhydramnios is associated with growth retardation as it may suggest chromosomal abnormality [57, 58].

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